II

110TH CONGRESS
2D SESSION

S. 2618

To amend the Public Health Service Act to provide for research with respect to various forms of muscular dystrophy, including Becker, congenital, distal, Duchenne, Emery-Dreifuss facioscapulohumeral, limb-girdle, myotonic, and oculopharyngeal muscular dystrophies.

IN THE SENATE OF THE UNITED STATES

FEBRUARY 8 (legislative day, FEBRUARY 6), 2008

Ms. KLOBUCHAR (for herself, Mr. ISAKSON, Mr. BROWN, Ms. COLLINS, Mr. COLEMAN, and Mr. HARKIN) introduced the following bill; which was read twice and referred to the Committee on Health, Education, Labor, and Pensions

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A BILL

To amend the Public Health Service Act to provide for research with respect to various forms of muscular dystrophy, including Becker, congenital, distal, Duchenne, Emery-Dreifuss facioscapulohumeral, limb-girdle, myotonic, and oculopharyngeal muscular dystrophies.

Be it enacted by the Senate and House of Representatives of the United States of America in Congress assembled,

SECTION 1. SHORT TITLE.

This Act may be cited as the ''Paul D. Wellstone Muscular Dystrophy Community Assistance, Research, and Education Amendments Act of 2008''.

SEC. 2. FINDINGS.

Congress makes the following findings:

(1) The muscular dystrophies are devastating diseases that have a significant impact on quality of life, not only for the individual who experiences its painful symptoms and resulting disability, but also for family members and caregivers.

(2) Duchenne muscular dystrophy (referred to in this section as ''DMD'') is the most common lethal genetic disorder of childhood worldwide, affecting approximately 1 in every 3,500 boys born each year around the globe. It is characterized by a rapidly progressive muscle weakness that almost always results in death from respiratory or cardiac failure, typically in the late teens or twenties.

(3) Myotonic muscular dystrophy is the second most prominent form of muscular dystrophy and the type most commonly found in adults, affecting an estimated 1 in 8,000 people. However, it can affect people of any age, from birth to old age. Described as the most variable disease known in medicine, it is multisystemic and can cause not only muscle atrophy and myotonia, but also serious cardiac, respiratory, endocrine, gastrointestinal, skeletal and central nervous system complications, as well as problems with the eyes, teeth, and hair. As it passes

from one generation to the next, it generally worsens with earlier onset. Congenital myotonic muscular dystrophy is the most severe form of myotonic muscular dystrophy affecting infants and causing severe cognitive delays. It often causes sudden death; however, others can live for many years with this slowly degenerative disorder.

(4) Facioscapulohumeral muscular dystrophy (referred to in this section as ''FSHD'') is the second most prevalent adult muscular dystrophy and the third most prevalent muscular dystrophy of men, women and children. It is inherited genetically and has an estimated incidence of 1 in 20,000 persons. Many leading FSHD scientists note that the prevalence may be 3 times higher due to undiagnosed and misdiagnosed cases. FSHD, affecting between 15,000 to 40,000 persons, causes a lifelong progressive and severe loss of all skeletal muscles gradually bringing weakness and reduced mobility. It is genetically transmitted to children, can occur spontaneously, and may affect entire families. Persons with FSHD may also experience hearing loss, vision problems, and respiratory insufficiency; some may become severely physically disabled and spend decades in a wheelchair and on a ventilator. FSHD is caused

by a novel epigenetic phenomenon not found in other forms of muscular dystrophy and is caused by a contraction of repetitive DNA previously thought to be ''junk DNA''. The unique epigenetic structure of FSHD is unprecedented in other muscular dystrophies and genetic disorders and demands novel approaches and new research groups. Understanding this mechanism will have great benefit to other areas of biomedical research including cancer and other disease of epigenetic origin.

(5) Congenital muscular dystrophies represent a group of distinct diseases, which begin at birth, with varying severity and involvement of both muscle strength and brain. These diseases often lead to premature infant death, or severely disabled young children who require 24-hour care given their developmental delay compounded by muscle weakness. Other children live to young adulthood and typically require the use of a wheelchair for mobility.

(6) Forms of muscular dystrophy affecting children and adults include Becker, congenital, distal, Duchenne, Emery-Dreifuss, facioscapulohumeral, limb-girdle, myotonic, and oculopharyngeal muscular dystrophies. The limb-girdle muscular dystrophies are of 15 known different types.

(7) Each of the muscular dystrophies, though distinct in progressivity and severity of symptoms, has a devastating impact on hundreds of thousands of children and adults throughout the United States and worldwide, as well as imposes severe physical and economic burdens on those affected. In many of the muscular dystrophies, there are associated medical problems arising from pulmonary issues, respiratory insufficiency, cardiomyopathy, which in many cases is the cause of death for persons with muscular dystrophy.

(8) In the 5 years since enactment of the Muscular Dystrophy Community Assistance, Research and Education Amendments of 2001 (the MD-CARE Act (Public Law 107–84)), and due directly to the momentum established by such Act, progress has been made in the battle against the muscular dystrophies.

(9) Investments made by the Federal Government as a result of the MD-CARE Act include the creation of the MD Coordinating Committee (MDCC), the development of the MDCC Action Plan, expansion of the National Institutes of Health (referred to in this section as the ''NIH'') research portfolios, establishment of 6 Paul D. Wellstone

Muscular Dystrophy Cooperative Research Centers, funding of a $15,400,000 National Institutes of Health U54 grant and others focused on DMD, development of the Muscular Dystrophy Surveillance, Tracking and Research Network (MD STARnet), and the launch of a comprehensive education and outreach initiative.

(10) In the past few years, the NIH program in translational research in muscular dystrophy has grown significantly and funded a number of large-scale projects to further the development of therapies for muscular dystrophy. As part of this program, the National Institute of Neurological Disorders and Stroke (NINDS) and the National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS) of the National Institutes of Health (NIH) awarded a $15,400,000, 5-year cooperative agreement to develop new small molecule drugs for the treatment of DMD and potentially other forms of muscular dystrophy as well. The project is a unique research collaboration between private, public, and nonprofit partners to build upon previous research and discovery work originally initiated by non-profit partners to identify new treatments for muscular dystrophy. Also through the translational

program, 3 other major cooperative agreements have been awarded for highly targeted therapy development projects in the muscular dystrophies.

(11) Due to the initiatives made possible through the MD-CARE Act, national nonprofit organizations have joined in model strategic collaborations with academic research institutions, public funders of muscular dystrophy research, and industry to expand investments in muscular dystrophy research activities and to create new platforms for translational research. These have led to the development of the first potential therapies for DMD, myotonic, facioscapulohumeral, limb-girdle, and other conditions that are proceeding through clinical trials.

(12) Advancements in care have helped prolong life and quality of life for patients with muscular dystrophy.

(13) Notwithstanding these promising developments, the majority of the directions envisioned by the Action Plan for the Muscular Dystrophies, developed pursuant to the MD-CARE Act, have not been realized. Where recent momentum has been achieved, its sustainability is fragile and directly dependent upon continued Federal support for the

early phase planning and programs created through the MD-CARE Act.

(14) There remains a shortage of qualified researchers in the field of muscular dystrophy research. Many family physicians and health care professionals still lack the knowledge and resources to detect and properly diagnose muscular dystrophy as early as possible, thus delaying management of symptoms in cases that go undetected or misdiagnosed.

(15) As new understandings of the genetic basis for disease and potential treatment has emerged, the public and health care communities are in urgent need of education and outreach to ensure competent, informed engagement in genetic testing and counseling and appropriate patient characterization so that patients are able to participate in new avenues of research and clinical trials.

(16) As basic research into the muscular dystrophies points the way to new therapeutic targets, there is an urgent need to support the clinical research infrastructure necessary to bring these therapeutic leads to human trials; these infrastructure needs include validated endpoints, current natural

history studies, biomarkers, clinical research networks, patient registries, and databases.

(17) In order to improve lives and develop effective treatments for individuals with muscular dystrophy, there must be improved communications and partnerships between patients, patient advocacy, researchers, and clinical care providers. To that end, renewed effort to work together by all parties is a critical element for successful outcomes in the years to come.

(18) Continued focus and investment are required to build on the current momentum, respond to public need, and ensure that federally funded research and other innovation is translated to therapeutic targets as quickly as possible.

SEC. 3. EXPANSION, INTENSIFICATION, AND COORDINATION OF ACTIVITIES OF NATIONAL INSTITUTES OF HEALTH WITH RESPECT TO RESEARCH ON MUSCULAR DYSTROPHY.

Section 404E of the Public Health Service Act (42 U.S.C. 283g) is amended—

(1) in subsection (a)(1), by inserting ''the National Heart, Lung, and Blood Institute,'' after ''Child Health and Human Development,'';

(2) in subsection (b)(1), by adding at the end the following: ''Such centers of excellence shall be known as the 'Paul D. Wellstone Muscular Dystrophy Cooperative Research Centers'.''; and

(3) by adding at the end the following:

''(h) CLINICAL RESEARCH.—The Coordinating Committee shall give special consideration to the urgent need to enhance the clinical research infrastructure required to test emerging therapies for the various forms of muscular dystrophy by prioritizing the achievement of those goals in the plan related to this topic.

''(i) AUTHORIZATION OF APPROPRIATIONS.—There is authorized to be appropriated to carry out this section, such sums as may be necessary for each of fiscal years 2008 through 2012.''.

SEC. 4. DEVELOPMENT AND EXPANSION OF ACTIVITIES OF CENTERS FOR DISEASE CONTROL AND PREVENTION WITH RESPECT TO EPIDEMIOLOGICAL RESEARCH ON MUSCULAR DYSTROPHY.

Section 317Q of the Public Health Service Act (42 U.S.C. 247b–18) is amended—

(1) by redesignating subsection (d) as subsection (f); and

(2) by inserting after subsection (c) the following:

''(d) DATA.—In carrying out this section, the Secretary shall ensure that any data on patients that is collected as part of the Muscular Dystrophy Surveillance, Tracking and Research Network (referred to in this section as the 'MD STARnet') under a grant under this section is regularly updated to reflect changes in patient condition over time, particularly with respect to any improvements realized through patient adherence to care considerations or utilization of a treatment or therapy.

''(e) REPORTS AND TRACKING.—

''(1) ANNUAL REPORT.—Not later than 18 months after the date of enactment of the Paul D. Wellstone Muscular Dystrophy Community Assistance, Research, and Education Amendments Act of 2008, and annually thereafter, the Director of the Centers for Disease Control and Prevention shall submit to the appropriate committees of Congress a report—

''(A) concerning the activities carried out by MD STARnet sites funded under this section during the year for which the report is prepared;

''(B) containing the data collected and findings derived from the MD STARnet sites each fiscal year (as funded under a grant under

this section during the periods of fiscal years 2008 through 2012); and

''(C) that every 2 years outlines prospective data collection objectives and strategies.

''(2) TRACKING HEALTH OUTCOMES.—The Director of the Centers for Disease Control and Prevention shall make publicly available prospective health outcome data on the health and survival of people with muscular dystrophy.''.

SEC. 5. INFORMATION AND EDUCATION.

Section 5 of the Muscular Dystrophy Community Assistance, Research and Education Amendments of 2001 (42 U.S.C. 247b–19) is amended—

(1) by redesignating subsection (c) as subsection (d); and

(2) by inserting after subsection (b), the following:

''(c) REQUIREMENTS OF THE CENTERS FOR DISEASE CONTROL AND PREVENTION.—In carrying out this section, the Director of the Centers for Disease Control and Prevention shall—

''(1) partner with leaders in the muscular dystrophy patient community; and

''(2) widely disseminate the Duchenne-Becker Muscular Dystrophy care considerations described in

section 904 as broadly as possible, including through partnership opportunities with the muscular dystrophy patient community.''.

SEC. 6. STANDARDS OF CARE.

Part A of title IX of the Public Health Service Act (42 U.S.C. 299 et seq.) is amended by adding at the end the following:

''SEC. 904. STANDARDS OF CARE RELATING TO MUSCULAR DYSTROPHY.

''The Director shall—

''(1) evaluate the available scientific evidence for the appropriate medical or patient organizations for purposes of the development and issuance of an initial set of care considerations for Duchenne-Becker Muscular Dystrophy and provide ongoing review and updates where appropriate; and

''(2) replicate the same systematic review methodology used to develop the Duchenne-Becker Muscular Dystrophy care considerations developed under paragraph (1) as a model for other muscular dystrophies.''.

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